Homocystinuria (HCY)

An amino acid disorder

What is it?

Homocystinuria (also known as HCY) is an inherited amino acid disorder. People with amino acid disorders, like HCY, cannot properly break down certain components of protein. This is because the body is lacking a specific chemical called an enzyme. Since the body cannot properly break down the protein, certain amino acids build up in the blood and urine and cause problems when a person eats normal amounts of protein.

What are the symptoms?

People with HCY may appear normal at birth. If untreated, a person with HCY will develop progressive vision problems, tall stature, slender build, scoliosis, mental retardation or developmental delay, seizures, and an increased risk of stroke can occur. Many symptoms of HCY can be prevented by immediate treatment and lifelong management. People with HCY typically receive follow-up care by a team of professionals that is experienced in treating people with metabolic disorders.

Inheritance and frequency

HCY is inherited in an autosomal recessive manner. This means that for a person to be affected with HCY, he or she must have inherited two non-working copies of the gene responsible for causing HCY. Usually, both parents of a person affected with an autosomal recessive disorder are unaffected because they are carriers. This means that they have one working copy of the gene, and one non-working copy of the gene. When both parents are carriers, there is a 1 in 4 (or 25%) chance that both parents will pass on the non working copies of their gene, causing the baby to have HCY. Typically, there is no family history of HCY in an affected person. HCY is a rare amino acid disorder; about 1 in 200,000 babies born have HCY.

How is it detected?

HCY may be detected through newborn screening. A recognizable pattern of elevated chemicals alerts the laboratory that a baby may be affected. Confirmation of newborn screening results is required to make a firm diagnosis. This is usually done by a physician that specializes in metabolic conditions, or a primary care physician.

How is it treated?

HCY is treated by eating a diet low in protein and drinking a special formula, and sometimes medication, as recommended by a genetic metabolic medical professional.

DISCLAIMER: This information is not intended to replace the advice of a genetic metabolic medical professional.

For more information:

Genetics Home Reference

Website: http://www.ghr.nlm.nih.gov

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4 Manor View Circle
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Toll Free Phone: 1-888-454-3383
Fax: (610) 993-0545
Email: email@savebabies.org
Website: http://www.savebabies.org

National Coalition of PKU and other allied disorders
http://www.pku-allieddisorders.org/